4p13 Deletion

Wolf-Hirschhorn Syndrome

Mode of Inheritance	• Primarily de novo deletions (87%) • Variable expressivity
Renal Phenotype	• Variable, including: vesicoureteral reflux, renal agenesis, cystic dysplasia, hypoplasia, horseshoe kidney, bladder exstrophy
Extra-renal Manifestations	• Failure to thrive • Microcephaly • Facial features: high forehead, hypertelorism, beaked nose, downturned mouth, micrognathia • Cleft lip or palate • Small bowel malrotation, reflux, absence of the gallbladder • Genital anomalies (e.g. hypospadias, cryptorchidism, clitoral aplasia or hyperplasia, absent uterus/vagina) • Scoliosis, polydactyly, fused vertebrae or ribs • Developmental delay and hypotonia • Hydrocephalus, corpus callosum abnormalities • Seizures
Pre-Transplant Management	• Screening and management of extra-renal manifestations
Transplant Considerations	• Careful screening of potential living related donors (inherited in an autosomal dominant manner)
Post-Transplant Management	• Low risk of disease recurrence

https://www.omim.org/entry/194190